Yi-Chung Lee

List of Publications by Year in descending order

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		182225	190340
127	3,525	30	53
papers	citations	h-index	g-index
130	130	130	6374
all docs	docs citations	times ranked	citing authors

#	Article	IF	CITATIONS
1	Incidental findings on brain magnetic resonance imaging: systematic review and meta-analysis. BMJ: British Medical Journal, 2009, 339, b3016-b3016.	2.4	634
2	Mutations in <i>KCND3</i> cause spinocerebellar ataxia type 22. Annals of Neurology, 2012, 72, 859-869.	2.8	138
3	Cutoff Scores of the Cognitive Abilities Screening Instrument, Chinese Version in Screening of Dementia. Dementia and Geriatric Cognitive Disorders, 2002, 14, 176-182.	0.7	129
4	The †hot cross bun†sign in the patients with spinocerebellar ataxia. European Journal of Neurology, 2009, 16, 513-516.	1.7	90
5	A Recurrent loss-of-function alanyl-tRNA synthetase (AARS ) mutation in patients with charcot-marie-tooth disease type 2N (CMT2N). Human Mutation, 2012, 33, 244-253.	1.1	90
6	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	1.5	86
7	The Mutational Spectrum in a Cohort of Charcot-Marie-Tooth Disease Type 2 among the Han Chinese in Taiwan. PLoS ONE, 2011, 6, e29393.	1.1	84
8	A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. Brain, 2017, 140, 1252-1266.	3.7	75
9	Population-specific spectrum of NOTCH3 mutations, MRI features and founder effect of CADASIL in Chinese. Journal of Neurology, 2009, 256, 249-255.	1.8	74
10	Characterization of CADASIL among the Han Chinese in Taiwan: Distinct Genotypic and Phenotypic Profiles. PLoS ONE, 2015, 10, e0136501.	1.1	73
11	Mutational analysis of MATR3 in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 2005.e1-2005.e4.	1.5	64
12	Mutations in ATP1A1 Cause Dominant Charcot-Marie-Tooth Type 2. American Journal of Human Genetics, 2018, 102, 505-514.	2.6	59
13	FUS, TARDBP, and SOD1 mutations in a Taiwanese cohort with familial ALS. Neurobiology of Aging, 2011, 32, 553.e13-553.e21.	1.5	57
14	Comparison of sensitivity of transcarpal median motor conduction velocity and conventional conduction techniques in electrodiagnosis of carpal tunnel syndrome. Clinical Neurophysiology, 2006, 117, 984-991.	0.7	55
15	Comparison of cerebellar ataxias: A threeâ€year prospective longitudinal assessment. Movement Disorders, 2011, 26, 2081-2087.	2.2	55
16	PRRT2 mutations lead to neuronal dysfunction and neurodevelopmental defects. Oncotarget, 2016, 7, 39184-39196.	0.8	55
17	A hexanucleotide repeat expansion in C9ORF72 causes familial and sporadic ALS in Taiwan. Neurobiology of Aging, 2012, 33, 2232.e11-2232.e18.	1.5	52
18	Visual cortex excitability and plasticity associated with remission from chronic to episodic migraine. Cephalalgia, 2012, 32, 537-543.	1.8	48

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19	PRRT2 Mutations in Paroxysmal Kinesigenic Dyskinesia with Infantile Convulsions in a Taiwanese Cohort. PLoS ONE, 2012, 7, e38543.	1.1	46
20	Exome Sequencing Identifies GNB4 Mutations as a Cause of Dominant Intermediate Charcot-Marie-Tooth Disease. American Journal of Human Genetics, 2013, 92, 422-430.	2.6	46
21	Extensive molecular genetic survey of Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2014, 35, 2423.e1-2423.e6.	1.5	46
22	Mutational analysis of TBK1 in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2016, 40, 191.e11-191.e16.	1.5	46
23	Selective Hypoperfusion of Anterior Cingulate Gyrus in Depressed AD Patients: A Brain SPECT Finding by Statistical Parametric Mapping. Dementia and Geriatric Cognitive Disorders, 2003, 16, 238-244.	0.7	41
24	Connectivity Features for Identifying Cognitive Impairment in Presymptomatic Carotid Stenosis. PLoS ONE, 2014, 9, e85441.	1.1	39
25	A novel <i>TFG</i> mutation causes Charcot-Marie-Tooth disease type 2 and impairs TFG function. Neurology, 2014, 83, 903-912.	1.5	39
26	Characterization of Heterozygous <i>HTRA1</i> Mutations in Taiwanese Patients With Cerebral Small Vessel Disease. Stroke, 2018, 49, 1593-1601.	1.0	39
27	Cognitive Reserve: A SPECT Study of 132 Alzheimer's Disease Patients with an Education Range of 0–19 Years. Dementia and Geriatric Cognitive Disorders, 2005, 20, 8-14.	0.7	38
28	Renal function is associated with 1-month and 1-year mortality in patients with ischemic stroke. Atherosclerosis, 2018, 269, 288-293.	0.4	38
29	Occipital lobe seizures related to marked elevation of hemoglobin A1C: Report of two cases. Seizure: the Journal of the British Epilepsy Association, 2010, 19, 359-362.	0.9	34
30	Spinocerebellar ataxia 35. Neurology, 2014, 83, 1554-1561.	1.5	33
31	Mutation spectrum of Charcotâ€Marieâ€Tooth disease among the Han Chinese in Taiwan. Annals of Clinical and Translational Neurology, 2019, 6, 1090-1101.	1.7	29
32	Clinical and genetic profiles of hereditary transthyretin amyloidosis in Taiwan. Annals of Clinical and Translational Neurology, 2019, 6, 913-922.	1.7	29
33	<i>NOTCH3</i> cysteine-altering variant is an important risk factor for stroke in the Taiwanese population. Neurology, 2020, 94, e87-e96.	1.5	28
34	Neuronal intranuclear inclusion disease in patients with adult-onset non-vascular leukoencephalopathy. Brain, 2022, 145, 3010-3021.	3.7	28
35	Transthyretin Ala97Ser in Chinese–Taiwanese patients with familial amyloid polyneuropathy: Genetic studies and phenotype expression. Journal of the Neurological Sciences, 2008, 267, 91-99.	0.3	26
36	A homozygous NOTCH3 mutation p.R544C and a heterozygous TREX1 variant p.C99MfsX3 in a family with hereditary small vessel disease of the brain. Journal of the Chinese Medical Association, 2013, 76, 319-324.	0.6	25

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37	Intracerebral Hemorrhage in Cerebral Autosomal Dominant Arteriopathy With Subcortical Infarcts and Leukoencephalopathy. Stroke, 2021, 52, 985-993.	1.0	25
38	Cerebral involvement in spinal and bulbar muscular atrophy (Kennedy's disease): A pilot study of PET. Journal of the Neurological Sciences, 2013, 335, 139-144.	0.3	24
39	Spinocerebellar ataxia type 36 in the Han Chinese. Neurology: Genetics, 2016, 2, e68.	0.9	24
40	GGC Repeat Expansion of <i>NOTCH2NLC</i> in Taiwanese Patients With Inherited Neuropathies. Neurology, 2022, 98, .	1.5	24
41	Statistical Parametric Mapping of Brain SPECT Perfusion Abnormalities in Patients with Alzheimer's Disease. European Neurology, 2003, 49, 142-145.	0.6	22
42	Two Novel De Novo GARS Mutations Cause Early-Onset Axonal Charcot-Marie-Tooth Disease. PLoS ONE, 2015, 10, e0133423.	1.1	22
43	Plasma amyloid assay as a pre-screening tool for amyloid positron emission tomography imaging in earlyÂstage Alzheimer's disease. Alzheimer's Research and Therapy, 2019, 11, 111.	3.0	21
44	Prolonged cortical relay time of long latency reflex and central motor conduction in patients with spinocerebellar ataxia type 6. Clinical Neurophysiology, 2003, 114, 458-462.	0.7	20
45	Transmissible spongiform encephalopathies with P102L mutation of PRNP manifesting different phenotypes: clinical, neuroimaging, and electrophysiological studies in Chinese kindred in Taiwan. Journal of Neurology, 2010, 257, 191-197.	1.8	20
46	Cerebral Microbleed Burdens in Specific Brain Regions Are Associated With Disease Severity of Cerebral Autosomal Dominant Arteriopathy With Subcortical Infarcts and Leukoencephalopathy. Journal of the American Heart Association, 2020, 9, e016233.	1.6	20
47	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy: Two novel mutations in the NOTCH3 gene in Chinese. Journal of the Neurological Sciences, 2006, 246, 111-115.	0.3	19
48	The remarkably variable expressivity of CADASIL: report of a minimally symptomatic man at an advanced age. Journal of Neurology, 2009, 256, 1026-1027.	1.8	19
49	Stenting versus Medical Treatment for Severe Symptomatic Intracranial Stenosis. American Journal of Neuroradiology, 2011, 32, 911-916.	1.2	19
50	Electrodiagnosis of Carpal Tunnel Syndrome: Which Transcarpal Conduction Technique Is Best?. Journal of Clinical Neurophysiology, 2009, 26, 366-371.	0.9	18
51	Clinical and Molecular Characterization of BSCL2 Mutations in a Taiwanese Cohort with Hereditary Neuropathy. PLoS ONE, 2016, 11, e0147677.	1.1	18
52	Charcot-Marie-Tooth disease. Current Treatment Options in Neurology, 2008, 10, 94-102.	0.7	17
53	Eye of the tiger-like MRI in parkinsonian variant of multiple system atrophy. Journal of Neural Transmission, 2009, 116, 861-866.	1.4	17
54	Blockade of soluble epoxide hydrolase attenuates post-ischemic neuronal hyperexcitation and confers resilience against stroke with TrkB activation. Scientific Reports, 2018, 8, 118.	1.6	17

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55	Common mitochondrial DNA and POLG1 mutations are rare in the Chinese patients with adult-onset ataxia on Taiwan. Journal of the Neurological Sciences, 2007, 254, 65-68.	0.3	16
56	Alteration of Proximal Conduction Velocity at Distal Nerve Injury in Carpal Tunnel Syndrome: Demyelinating Versus Axonal Change. Journal of Clinical Neurophysiology, 2008, 25, 161-166.	0.9	16
57	Cellular characterization of MPZ mutations presenting with diverse clinical phenotypes. Journal of Neurology, 2010, 257, 1661-1668.	1.8	16
58	The interaction of serum testosterone levels and androgen receptor CAG repeat polymorphism on the risk of erectile dysfunction in aging Taiwanese men. Andrology, 2015, 3, 902-908.	1.9	16
59	Clinical and biophysical characterization of 19 <i>GJB1</i> mutations. Annals of Clinical and Translational Neurology, 2016, 3, 854-865.	1.7	16
60	A novel <i><scp>DNAJB6</scp></i> mutation causes dominantly inherited distalâ€onset myopathy and compromises <scp>DNAJB6</scp> function. Clinical Genetics, 2017, 92, 150-157.	1.0	16
61	Investigating CYP2C19 loss-of-function allele statuses and their association with stroke of different etiologies in a Taiwanese population. Journal of the Chinese Medical Association, 2019, 82, 469-472.	0.6	16
62	Mitochondrial DNA damage in spinal and bulbar muscular atrophy patients and carriers. Clinica Chimica Acta, 2010, 411, 626-630.	0.5	14
63	Validating a rapid, real-time, PCR-based direct mutation detection assay for preimplantation genetic diagnosis. Gene, 2014, 548, 299-305.	1.0	14
64	Comparable progression of spinocerebellar ataxias between Caucasians and Chinese. Parkinsonism and Related Disorders, 2019, 62, 156-162.	1.1	14
65	Epileptic seizures in a patient by immersing his right hand into hot water. Seizure: the Journal of the British Epilepsy Association, 2000, 9, 605-607.	0.9	13
66	Incidental Findings on Brain MRI. New England Journal of Medicine, 2008, 358, 853-855.	13.9	13
67	FGF21 in ataxia patients with spinocerebellar atrophy and mitochondrial disease. Clinica Chimica Acta, 2012, 414, 225-227.	0.5	13
68	What we have learned from the next-generation sequencing: Contributions to the genetic diagnoses and understanding of pathomechanisms of neurodegenerative diseases. Journal of Neurogenetics, 2015, 29, 103-112.	0.6	13
69	Investigating CCNF mutations in a Taiwanese cohort with amyotrophic lateral sclerosis. Neurobiology of Aging, 2018, 62, 243.e1-243.e6.	1.5	13
70	Rapid progressive ALS in a patient with a <i>DNAJC7</i> loss-of-function mutation. Neurology: Genetics, 2020, 6, e503.	0.9	13
71	Clinical and Genetic Characterization of Autosomal Recessive Spinocerebellar Ataxia Type 16 (SCAR16) in Taiwan. Cerebellum, 2020, 19, 544-549.	1.4	13
72	Mutational analysis of ITPR1 in a Taiwanese cohort with cerebellar ataxias. PLoS ONE, 2017, 12, e0187503.	1.1	13

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73	An environmental decisionâ€support system for the management of water pollution in a tidal river network. International Journal of Geographical Information Science, 2005, 19, 483-500.	2.2	12
74	<i>MPZ</i> mutation G123S characterization. Neurology, 2008, 70, 273-277.	1.5	12
75	Genetic analysis of ANXA11 variants in a Han Chinese cohort with amyotrophic lateral sclerosis in Taiwan. Neurobiology of Aging, 2018, 72, 188.e1-188.e2.	1.5	12
76	Clinical characteristics of Taiwanese patients with Hereditary spastic paraplegia type 5. Annals of Clinical and Translational Neurology, 2020, 7, 486-496.	1.7	12
77	Intronic CArG Box Regulates Cysteine-Rich Protein 2 Expression in the Adult but Not in Developing Vasculature. Arteriosclerosis, Thrombosis, and Vascular Biology, 2010, 30, 835-842.	1.1	11
78	SCA31 is rare in the Chinese population on Taiwan. Neurobiology of Aging, 2012, 33, 426.e23-426.e24.	1.5	11
79	Allergic rhinitis and risk of erectile dysfunction – a nationwide populationâ€based study. Allergy: European Journal of Allergy and Clinical Immunology, 2013, 68, 440-445.	2.7	11
80	C9ORF72 repeat expansion is not a significant cause of late onset cerebellar ataxia syndrome. Journal of the Neurological Sciences, 2014, 347, 322-324.	0.3	11
81	Unmasking adrenoleukodystrophy in a cohort of cerebellar ataxia. PLoS ONE, 2017, 12, e0177296.	1.1	11
82	Prolonged central motor conduction time of lower limb muscle in spinocerebellar ataxia 6. Journal of Clinical Neuroscience, 2004, 11, 381-383.	0.8	10
83	The role of forearm mixed nerve conduction study in the evaluation of proximal conduction slowing in carpal tunnel syndrome. Clinical Neurophysiology, 2008, 119, 2800-2803.	0.7	10
84	Clinical and Molecular Characterization of PMP22 point mutations in Taiwanese patients with Inherited Neuropathy. Scientific Reports, 2017, 7, 15363.	1.6	10
85	Median nerve motor conduction velocity is concordant with myelin protein zero gene mutation. Journal of Neurology, 2005, 252, 151-155.	1.8	9
86	Bromism caused by mix-formulated analgesic injectables. Human and Experimental Toxicology, 2007, 26, 971-973.	1.1	9
87	Mutational analysis of the 5′ non-coding region of GJB1 in a Taiwanese cohort with Charcot–Marie–Tooth neuropathy. Journal of the Neurological Sciences, 2013, 332, 51-55.	0.3	9
88	Analysis of NOTCH2NLC GGC repeat expansion in Taiwanese patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2021, 108, 210-212.	1.5	9
89	Clinical and genetic characterization of adultâ€onset leukoencephalopathy caused by <i>CSF1R</i> mutations. Annals of Clinical and Translational Neurology, 2021, 8, 2121-2131.	1.7	9
90	The Real Role of Forearm Mixed Nerve Conduction Velocity in the Assessment of Proximal Forearm Conduction Slowing in Carpal Tunnel Syndrome. Journal of Clinical Neurophysiology, 2008, 25, 373-377.	0.9	8

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91	Clinical characterization and genetic analysis of a possible novel type of dominant Charcot-Marie-Tooth disease. Neuromuscular Disorders, 2010, 20, 534-539.	0.3	8
92	Acute simultaneous multiple lacunar infarcts as the initial presentation of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Journal of the Chinese Medical Association, 2015, 78, 424-426.	0.6	8
93	Investigating PUM1 mutations in a Taiwanese cohort with cerebellar ataxia. Parkinsonism and Related Disorders, 2019, 66, 220-223.	1.1	8
94	Handâ€onset weakness is a common feature of ALS patients with a NEK1 lossâ€ofâ€function variant. Annals of Clinical and Translational Neurology, 2020, 7, 965-971.	1.7	8
95	Treatment response, risk of relapse and clinical characteristics of Taiwanese patients with neuromyelitis optica spectrum disorder. Journal of the Formosan Medical Association, 2022, 121, 1647-1656.	0.8	8
96	Myelin protein zero gene mutations in Taiwanese patients with Charcot–Marie–Tooth disease type 1. Journal of the Neurological Sciences, 2004, 219, 95-100.	0.3	7
97	Clinical and genetic characterization of hereditary spastic paraplegia type 3A in Taiwan. Parkinsonism and Related Disorders, 2021, 87, 87-91.	1.1	7
98	A <scp>PIAS1</scp> Protective Variant <scp>S510G</scp> Delays <scp>polyQ</scp> Disease Onset by Modifying Protein Homeostasis. Movement Disorders, 2021, , .	2.2	7
99	Wernicke's Encephalopathy in a Patient with Multiple Symmetrical Lipomatosis and the A8344G Mutation of Mitochondrial DNA. European Neurology, 2002, 47, 126-128.	0.6	6
100	Motor neuron disease-like syndrome secondary to trapped fourth ventricle and obstruction of cerebrospinal fluid pathway. Clinical Neurology and Neurosurgery, 2007, 109, 383-387.	0.6	6
101	Cholesterol Levels Are Associated with 30-day Mortality from Ischemic Stroke in Dialysis Patients. Journal of Stroke and Cerebrovascular Diseases, 2017, 26, 1349-1356.	0.7	6
102	Longitudinal Cerebral Perfusion Decrease in Mild Alzheimer's Disease Revealed by SPECT with Statistical Parametric Mapping Method. European Neurology, 2004, 52, 42-49.	0.6	5
103	Clinical and cellular characterization of two novel MPZ mutations, p.I135M and p.Q187PfsX63. Clinical Neurology and Neurosurgery, 2012, 114, 124-129.	0.6	5
104	Electrophysiological characterization of Charcot–Marie–Tooth disease type 1A in Taiwan. Journal of the Chinese Medical Association, 2012, 75, 197-202.	0.6	5
105	Preimplantation Genetic Diagnosis of Neurodegenerative Diseases: Review of Methodologies and Report of Our Experience as a Regional Reference Laboratory. Diagnostics, 2019, 9, 44.	1.3	5
106	Expanding the phenotype of AFG3L2 mutations: Late-onset autosomal recessive spinocerebellar ataxia. Journal of the Neurological Sciences, 2021, 428, 117600.	0.3	5
107	Gabapentin for complex regional pain syndrome in Machado-Joseph disease: a case report. Journal of Medical Case Reports, 2011, 5, 268.	0.4	4
108	A case of GNE myopathy mimicking hereditary motor neuropathy. European Journal of Neurology, 2020, 27, 2389-2391.	1.7	4

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109	Mutation screening and association analysis of <i>NOTCH3</i> p.R544C in patients with migraine with or without aura. Cephalalgia, 2022, 42, 888-898.	1.8	4
110	Spastic paraparesis as a manifestation of metabolic vitamin B12 deficiency. Journal of Neurology, 2005, 252, 1125-1126.	1.8	3
111	Mutational analysis of <i>CCM1</i> , <i>CCM2</i> and <i>CCM3</i> in a Han Chinese cohort with multiple cerebral cavernous malformations in Taiwan. Clinical Genetics, 2018, 94, 389-390.	1.0	3
112	Investigating ZFYVE26 mutations in a Taiwanese cohort with hereditary spastic paraplegia. Journal of the Formosan Medical Association, 2022, 121, 126-133.	0.8	3
113	Investigating TBP CAG/CAA trinucleotide repeat expansions in a Taiwanese cohort with ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 442-447.	1.1	3
114	Gain of C-Ala enables AlaRS to target the L-shaped tRNAAla. Nucleic Acids Research, 2022, 50, 2190-2200.	6.5	3
115	Fatigue in Colchicine Myopathy: A Study of Transcranial Magnetic Stimulation. Journal of the Chinese Medical Association, 2010, 73, 623-627.	0.6	2
116	Investigation for SPTLC1 mutations in a Taiwanese cohort with hereditary neuropathies. Journal of the Neurological Sciences, 2017, 381, 463-464.	0.3	2
117	Mitochondrial DNA m.3243A>G mutation rarely causes CADASIL-like phenotype. Neurobiology of Aging, 2021, 97, 145.e5-145.e6.	1.5	2
118	Genetic and Functional Analysis of Glycosyltransferase 8 Domain–Containing Protein 1 in Taiwanese Patients With Amyotrophic Lateral Sclerosis. Neurology: Genetics, 2021, 7, e627.	0.9	2
119	Assessing the NOTCH2NLC GGC repeat expansion in Taiwanese patients with hereditary spastic paraplegia. Parkinsonism and Related Disorders, 2022, 96, 43-44.	1.1	2
120	Coexistence of Charcot Marie Tooth disease type 1A and diabetes in Taiwan: A clinicopathological study. Journal of the Neurological Sciences, 2015, 358, 213-220.	0.3	1
121	Investigating ABCD1 mutations in a Taiwanese cohort with hereditary spastic paraplegia phenotype. Parkinsonism and Related Disorders, 2021, 92, 7-12.	1.1	1
122	Gene symbol: NOTCH3. Disease: cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. Human Genetics, 2005, 116, 242.	1.8	1
123	Reply: A novel WARS mutation causes distal hereditary motor neuropathy in a Chinese family. Brain, 2019, 142, e50-e50.	3.7	0
124	Prealbumin Is Associated with the Weaning Outcome for Patients on Prolonged Mechanical Ventilation. , 2020, , .		0
125	Reply: MELAS can be delineated from CADASIL by genotype and phenotype. Neurobiology of Aging, 2021, 99, 104.	1.5	0
126	Reply to: Adultâ€onset leukoencephalopathy caused by CSF1R mutations: Is all that glitters gold?. Annals of Clinical and Translational Neurology, 2022, 9, 101-102.	1.7	0

YI-CHUNG LEE

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127	Transient Postictal Hyperglycemia as a Diagnostic Clue of Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episodes Acta Neurologica Taiwanica, 2022, 31(2), 79-83.	0.3	O